



GLRA1 gene

glycine receptor alpha 1

Normal Function

The *GLRA1* gene provides instructions for making one part, the alpha (α)1 subunit, of the glycine receptor protein. The glycine receptor is most abundant in nerve cells (neurons) in the spinal cord and the part of the brain that is connected to the spinal cord (the brainstem). The glycine receptor is made up of five subunits: two α 1 subunits and three beta (β) subunits. The β subunit is produced from a different gene.

Receptor proteins have specific sites into which certain other molecules, called ligands, fit like keys into locks. Together, ligands and their receptors trigger signals that affect cell development and function. The ligand for the glycine receptor is the amino acid glycine. This molecule acts as a neurotransmitter, which is a chemical messenger that transmits signals in the nervous system.

When glycine attaches (binds) to the glycine receptor, the receptor opens to allow negatively charged chlorine atoms (chloride ions) to enter the cell. This influx of chloride ions reduces the cell's ability to transmit signals to other cells. Because they stop (inhibit) signaling, glycine receptors are known as inhibitory receptors.

Health Conditions Related to Genetic Changes

hereditary hyperekplexia

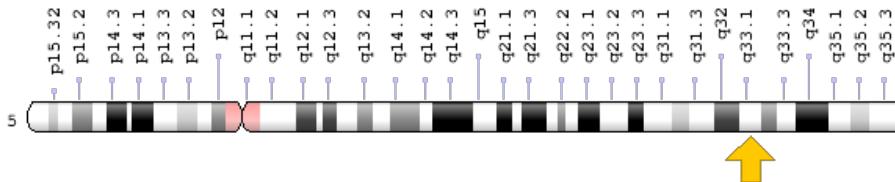
At least 29 mutations in the *GLRA1* gene have been found to cause hereditary hyperekplexia. Most of these mutations change single amino acids in the α 1 subunit of the glycine receptor protein. The most common mutation replaces the amino acid arginine with the amino acid leucine at protein position 271 (written as Arg271Leu or R271L).

GLRA1 gene mutations that cause hereditary hyperekplexia impair the ability of the glycine receptor protein to respond to the ligand glycine. Some *GLRA1* gene mutations alter the structure of the glycine receptor, which prevents the receptor from opening. Other mutations prevent the receptor from reaching the cell membrane. When the glycine receptor is nonfunctional or missing, chloride ions cannot enter the cell, and cell signaling is increased. This overactive cell signaling in the spinal cord and brainstem is thought to cause the abnormal muscle movements, exaggerated startle reaction, and other symptoms of hereditary hyperekplexia.

Chromosomal Location

Cytogenetic Location: 5q33.1, which is the long (q) arm of chromosome 5 at position 33.1

Molecular Location: base pairs 151,820,798 to 151,924,836 on chromosome 5 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- GLRA1_HUMAN
- glycine receptor, alpha 1
- glycine receptor, alpha 1 isoform 1 precursor
- glycine receptor, alpha 1 isoform 2 precursor
- STHE

Additional Information & Resources

Educational Resources

- Basic Neurochemistry (sixth edition, 1999): Glycine Receptors
<https://www.ncbi.nlm.nih.gov/books/NBK28003/>
- Washington University, St. Louis: Neuromuscular Disease Center
<http://neuromuscular.wustl.edu/mother/activity.html#hyperekplexia>

GeneReviews

- Hyperekplexia
<https://www.ncbi.nlm.nih.gov/books/NBK1260>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28GLRA1%5BTIAB%5D%29+OR+%28glycine+receptor+alpha+1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- GLYCINE RECEPTOR, ALPHA-1 SUBUNIT
<http://omim.org/entry/138491>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_GLRA1.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=GLRA1%5Bgene%5D>
- HGNC Gene Family: Glycine receptors
<http://www.genenames.org/cgi-bin/genefamilies/set/868>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=4326
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/2741>
- UniProt
<http://www.uniprot.org/uniprot/P23415>

Sources for This Summary

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